



The PerkinElmer Omics Laboratory

Todd Smith

PerkinElmer | Geospiza

SFAF June 5, 2012

PerkinElmer: dedicated to improving human & environmental health...



75 Years of Analytical Chemistry and Instrumentation Experience



Cellular research



Cancer research



GPCRs



Protein:protein



Epigenetics



Kinases



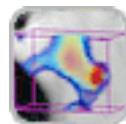
Immuno assays



Label free assays



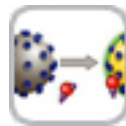
High throughput screening



In Vivo



Screening technologies



SPA "generic" assays



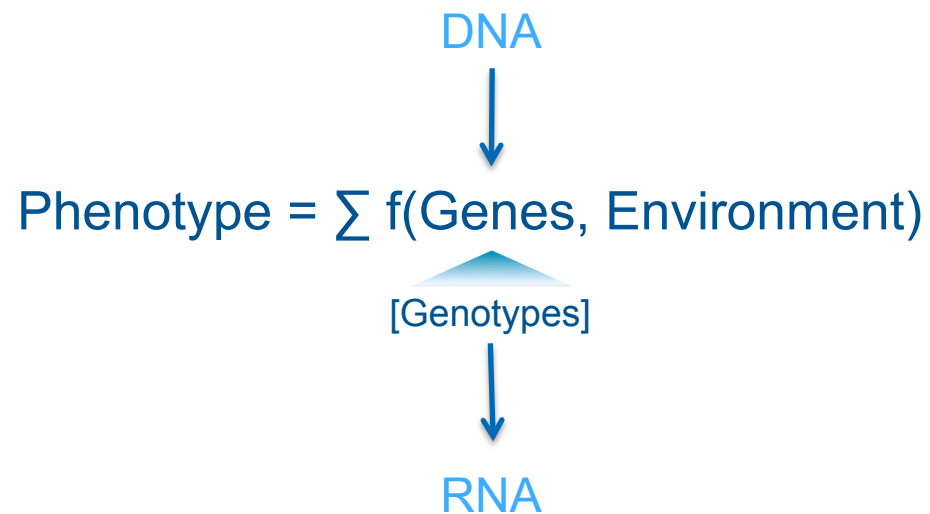
Biotherapeutics



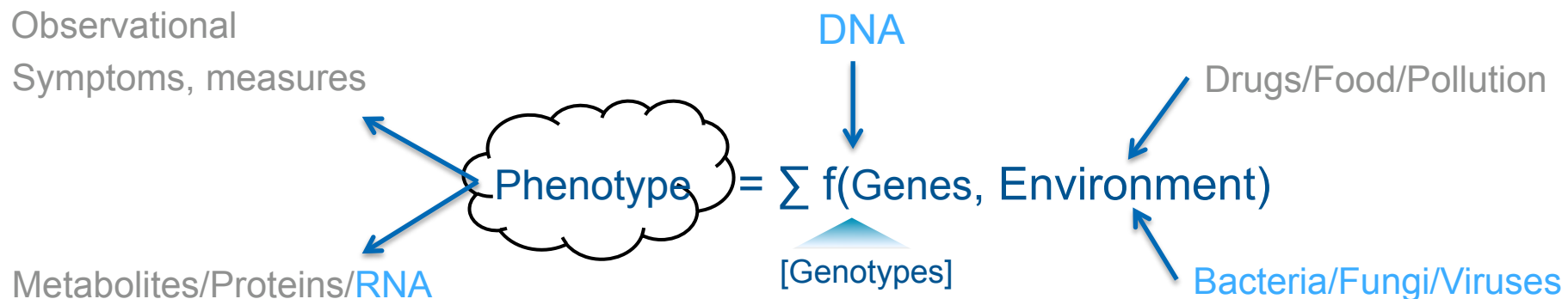
Radioligand binding



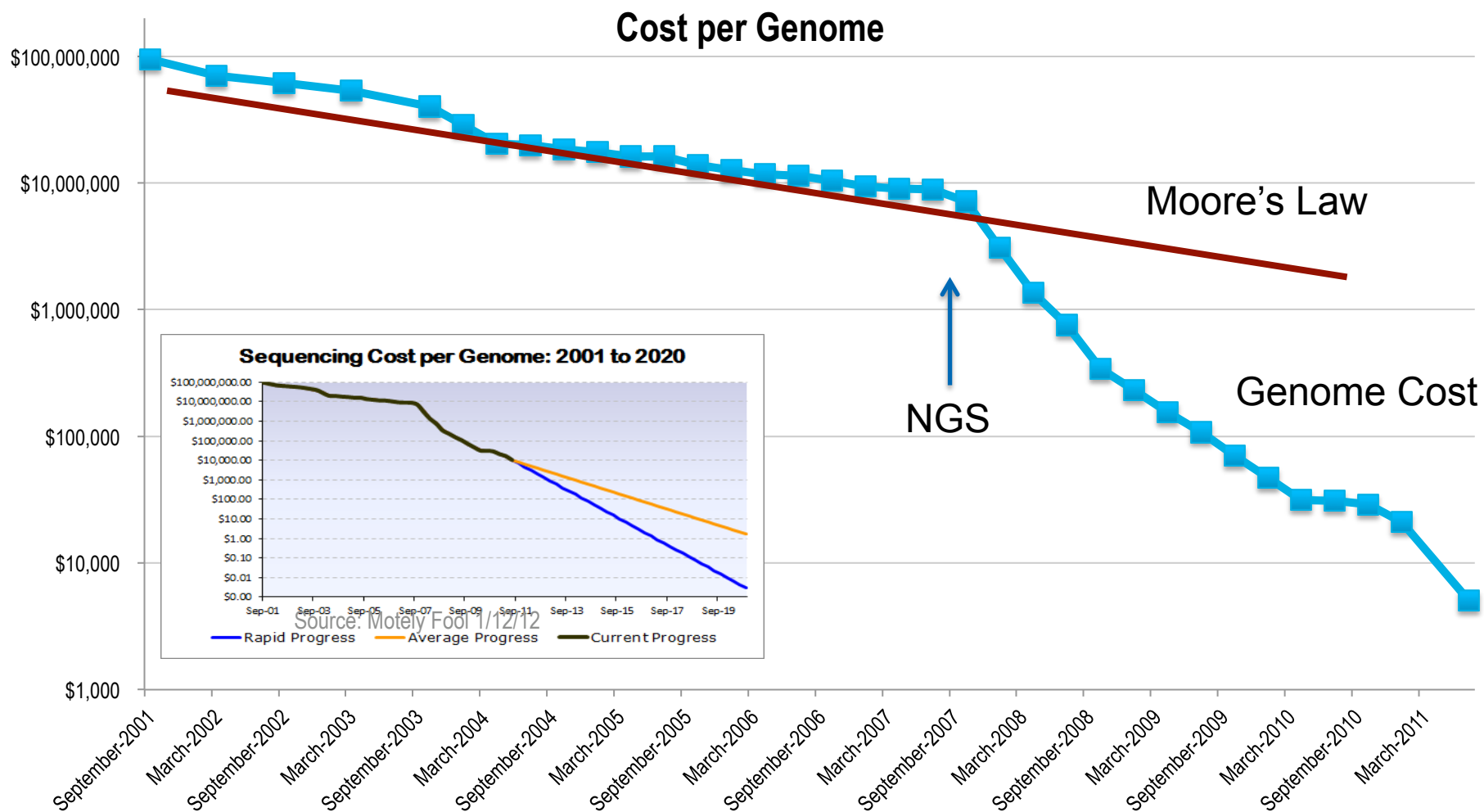
Laboratory automation



The Phenotype/Genotype Relationship: It's Complicated

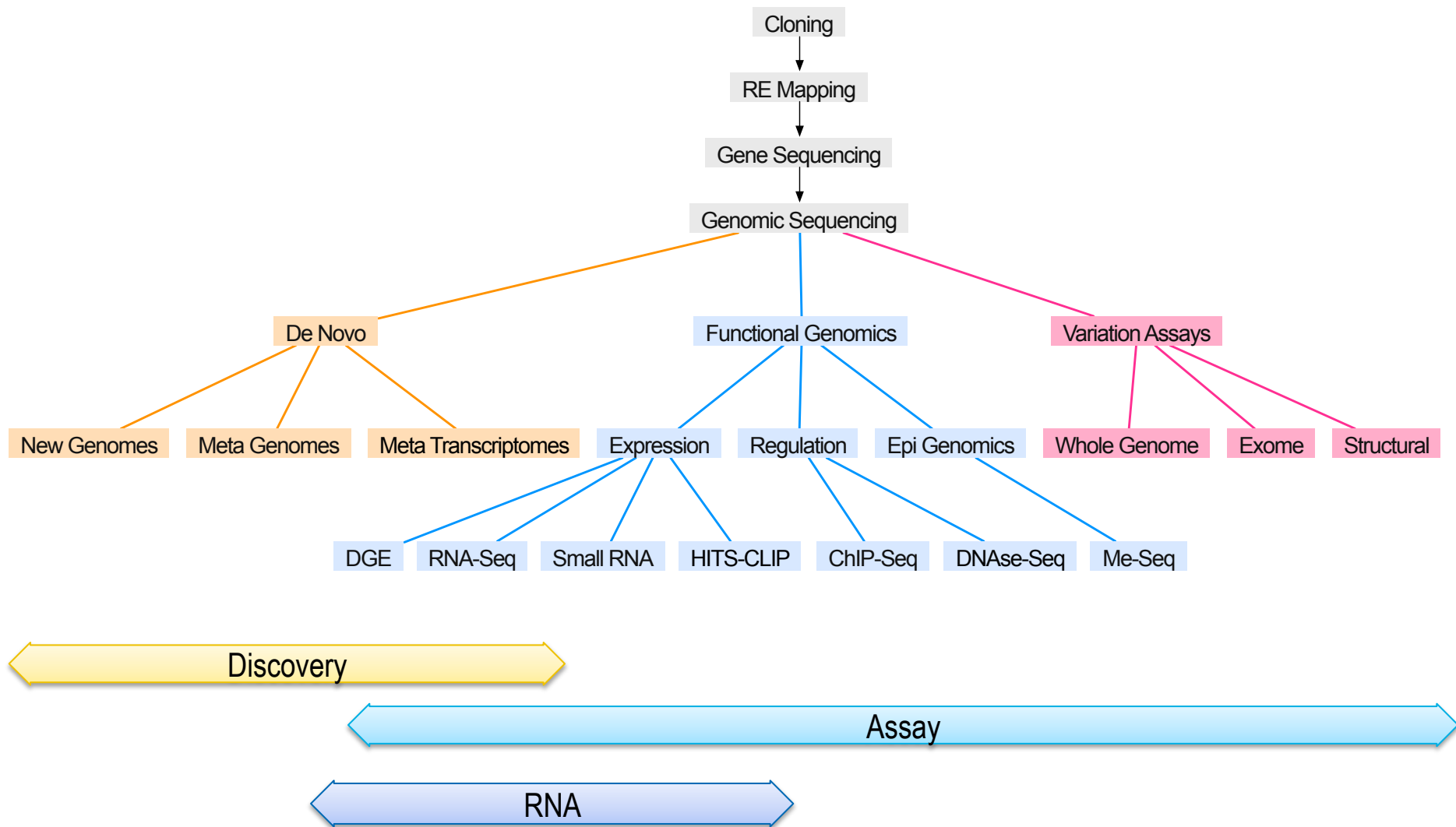


Increasing Access to Cheap Data



Source: NHGRI

Genomics Genealogy





January 2011

- Geospiza is selected to provide LIMS and analysis support PerkinElmer's DNA services



May 2011

- PerkinElmer acquires Geospiza

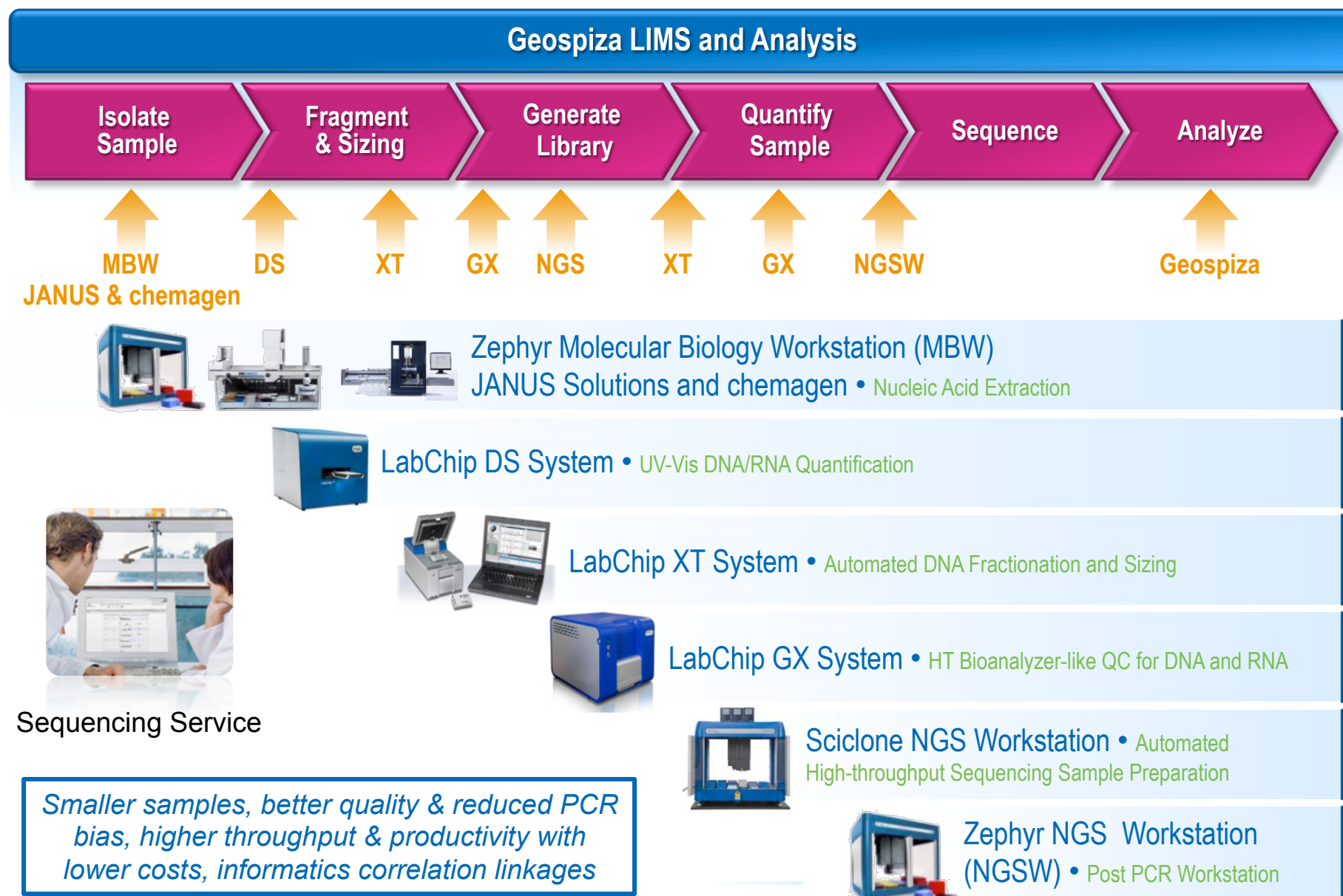


November 2011

- PerkinElmer acquires Caliper Life Sciences

Two bottlenecks: Sample Prep and Informatics

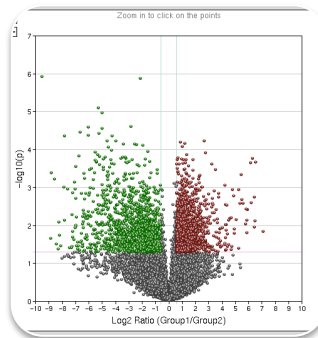
Omics Applications Demand Automation and Informatics





GeneSifter® Lab Edition

- Comprehensive web-based LIMS for genomics
- Scalable, flexible, adaptable
- Unique in accounting, and linkage with data analysis



GeneSifter® Analysis Edition

- Comprehensive web-based data analysis
- From microarrays to NGS
- Unique in completeness for creating new knowledge

[illegible]

A Better Way



Single sample

[Main \(login: eric_test\)](#) > [Inventories](#) > [Next Gen Files](#) > [Job Details](#) > [Result Files](#)

>> Analysis Result File: Variant Report (Data Files: SRR018257_1.fastq / SRR018257_2.fastq) **Number of Results: 12349**

Region + Show: [1 - 20] [21 - 40] Last

Chromosome	Position	dbSNP	Variant Type	Change	Description	Read Depth	Score	Gene ID	Region	Function	Change	CA	PGKB
chr1	887,188	rs49704441	snv	G<=>C	C3	3	36	KLHL17	exon	synonymous	-	-	-
chr1	924,935	no	insertion	*/+CT	+CT+1, *+2	3	29	HES4	exon	exon	-	-	-
chr1	968,949	no	deletion	*/-TG	-TG+1, *+2	3	29	AGR1	exon	exon	-	-	-
chr1	971,734	rs2465128	snv	A<=>G	C1, G8	9	51	AGR1	exon	synonymous	-	-	-
chr1	972,857	rs10267	snv	T<=>C	C6	6	45	AGR1	exon	synonymous	-	-	-
chr1	1,142,944	no	deletion	*/-GGG	-GGG+1, *+2	3	29	SDF4	exon	exon	-	-	-
chr1	1,237,357	rs12103	snv	T<=>C	C3	9	54	CPSF3L	exon	synonymous	-	-	-
chr1	1,238,789	no	snv	T<=>C	C2, T1	3	33	CPSF3L	exon	missense	G<=>R	-	-
chr1	1,244,704	rs10907179	snv	C<=>G	G8	8	51	CPSF3L	exon	synonymous	-	-	-
chr1	1,252,829	rs307349	snv	C<=>T	T5	5	35	GLTPD1	exon	synonymous	-	-	-
chr1	1,267,608	no	snv	A<=>G	A1, G2	3	33	DVL1	exon	missense	F<=>S	-	-
chr1	1,312,728	no	snv	A<=>C	A1, C2, G1	4	28	CONL2	exon	synonymous	-	-	-
chr1	1,315,473	no	snv	C<=>G	C1, G2	3	33	CONL2	exon	missense	V<=>L	-	-
chr1	1,316,019	no	snv	C<=>S	A2, C18, G5	26	50	CONL2	exon	missense	R<=>P	-	-
chr1	1,324,299	no	snv	A<=>G	A1, G3	4	24	CONL2	exon	missense	L<=>P	-	-
chr1	1,469,196	rs7553	snv	A<=>C	A12, G15	27	64	SSU72	exon	synonymous	-	-	-
chr1	1,640,667	rs3200657	snv	T<=>C	C3, T1	4	21	CDCL1	exon	synonymous	-	-	-
chr1	1,653,769	no	deletion	*/-CAT	-CAT+1, *+2	3	29	SLC35E2	exon	exon	-	-	-
chr1	1,677,568	no	snv	A<=>R	A4, G3	7	32	NADK	exon	missense	S<=>P	-	-
chr1	2,115,999	rs6662296	snv	C<=>G	G6	6	29	C1orf86	exon	missense	R<=>P	-	-

Region + Show: [1 - 20] [21 - 40] Last

Many samples

Analysis Result File: Multi-Sample Variant Report (SQLite)

Main (login, edit, test)

> Data

> Files

> Job Details

> Result File

Number of Results: 1228/9

Type

snv

+

OR

Region

CDS

-

Unique Variants

Show: 20

Color Variants:

Search

Reset

Expand All Collapse All

NEGO Export

Chrom	Position	Gene	Region	Type	dbSNP	Exon Pos	Low Cov	Pip	Ref.	N11: Var	C11: Var	N13: Var	C13: Var	N15: Var	C15: Var	N19: Var	C19: Var	
chr1	902,994	AGRN	CDS	snv	rs10287	0	0	T	T	C	C	M	C	C	C	C	C	C
chr1	1,254,841	CPH3L	CDS	snv	rs10897179	0	0	C	C	A	A	A	A	A	A	A	A	A
chr1	8,704,635	DNAJC11	CDS	snv	rs200458	0	0	A	A	T	T	T	T	T	T	T	T	T
chr1	16,133,987	NG2,25371	CDS	snv	rs7417355	0	0	T	T	Y	Y	Y	Y	Y	Y	Y	Y	Y
chr1	26,609,259	UBI011	CDS	snv	rs7222905	0	0	C	C	Y	Y	Y	Y	Y	Y	Y	Y	Y
chr1	41,204,589	MPYC	CDS	snv	rs1057925	0	0	C	C	Y	Y	Y	Y	Y	Y	Y	Y	Y
chr1	53,544,439	POGN	CDS	snv	rs128388	0	0	G	G	A	A	A	A	A	A	A	A	A
chr1	151,006,559	PRUNE	CDS	snv	rs3739476	0	0	C	C	M	A	A	A	A	A	A	A	A
chr2	29,135,468	VCOR2	CDS	snv	rs6715566	0	0	C	C	A	A	A	A	A	A	A	A	A
chr3	4,866,188	JPR1	CDS	snv	rs719331	0	0	T	T	C	C	C	C	C	C	C	C	C
chr3	47,452,087	PTPNC3	CDS	snv	rs905889	0	0	0	0	G	A	A	A	A	A	A	A	A
chr3	69,230,801	FRMD4B	CDS	snv	rs9833055	0	0	G	G	A	A	A	A	A	A	A	A	A
chr4	40,438,516	RBH47	CDS	snv	rs2307448	0	0	A	A	C	C	C	C	C	C	C	C	C
chr5	180,278,393	ZFP62	CDS	snv	rs765441	0	0	C	C	T	T	T	T	T	T	T	T	T
chr6	29,911,228	HLA-A	CDS	snv	rs112069136, rs70299908, rs76248018, rs7527524, rs6258983	0	0	A	A	C	C	M	Y	Y	M	M	M	M
chr6	32,625,690	PSMB9	CDS	snv	rs1140460706, rs17587, rs17432227	0	0	G	G	R	A	A	A	A	R	R	R	R
chr7	44,986,497	DENL	CDS	snv	-	0	0	C	C	C	C	C	C	C	C	C	C	C
chr7	75,659,815	STYXL1	CDS	snv	rs1044483	0	0	T	T	C	Y	C	C	C	C	C	C	C
chr7	100,548,873	MUC3A	CDS	snv	rs74103666	0	0	G	G	A	A	A	A	R	R	R	R	A
chr7	196,555,133	MUC3A	CDS	snv	rs74197937	0	0	A	A	R	R	R	R	R	R	R	R	R

Type

snv

+

OR

Region

CDS

-

Unique Variants

Show: 20

Color Variants:

Search

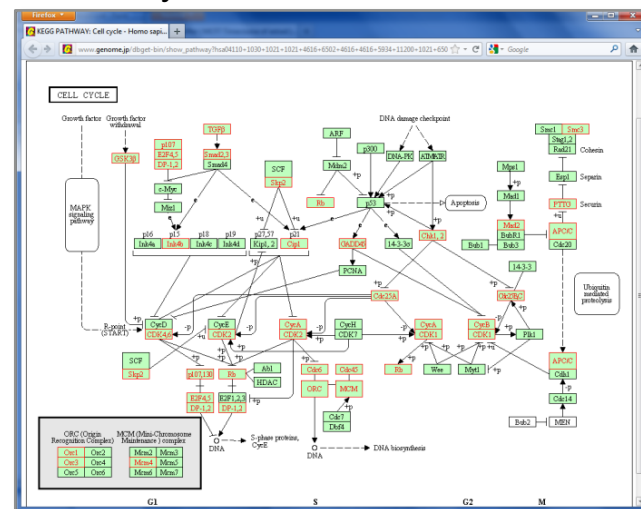
Reset

Expand All Collapse All

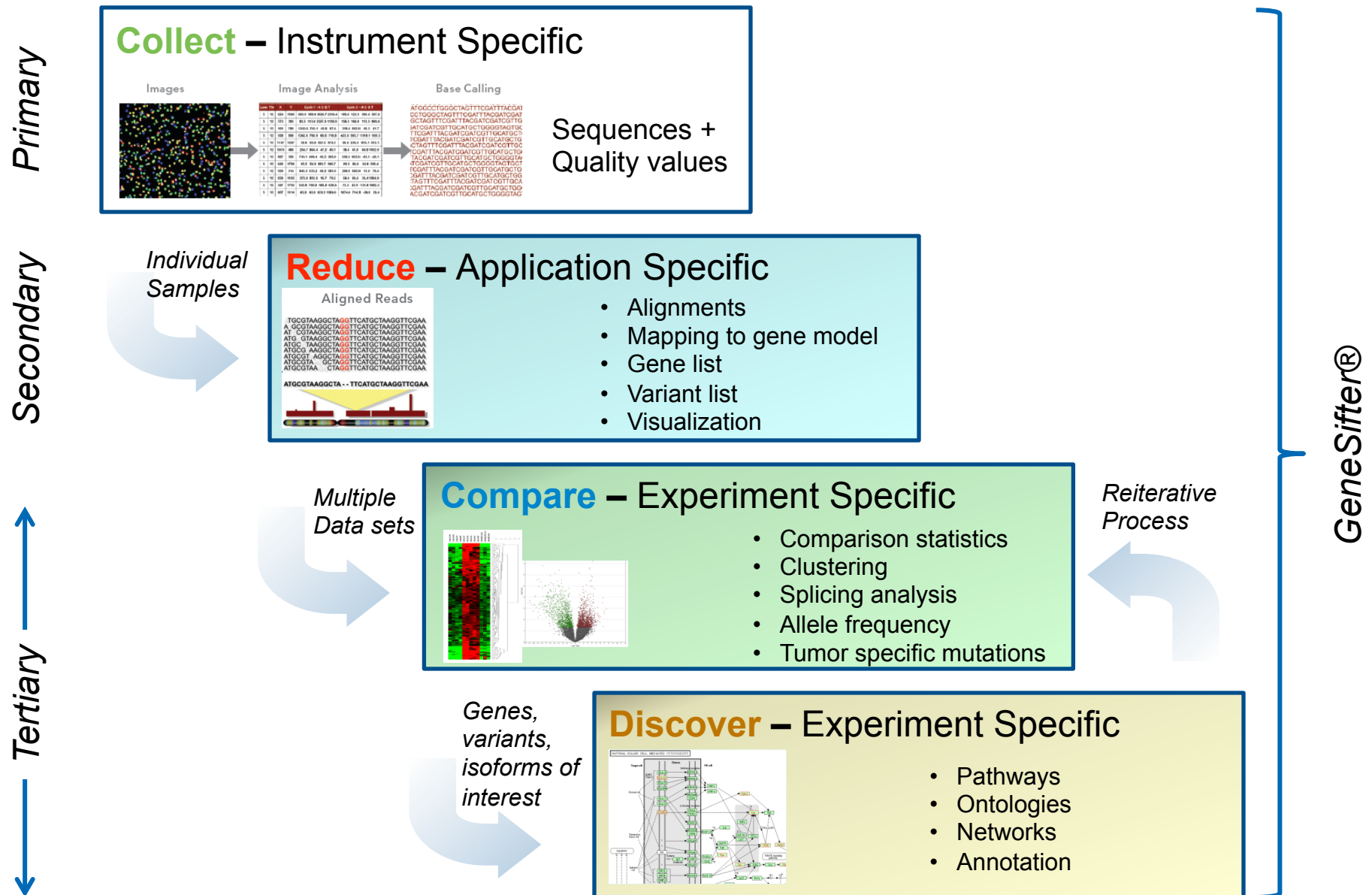
Base level



Pathways



Geospiza® NGS Data Analysis Workflow



What do you See in Your Experiment?

Pairwise Analysis: Prostate Cancer GSE22260 [Reports: Ontology | K]

Group 1

Conditions: Normal

Experiments: 87527, 87534

Significance: 1.5 - 5, edgeR, Benjamini and Hochberg

Normalization: Mapped reads

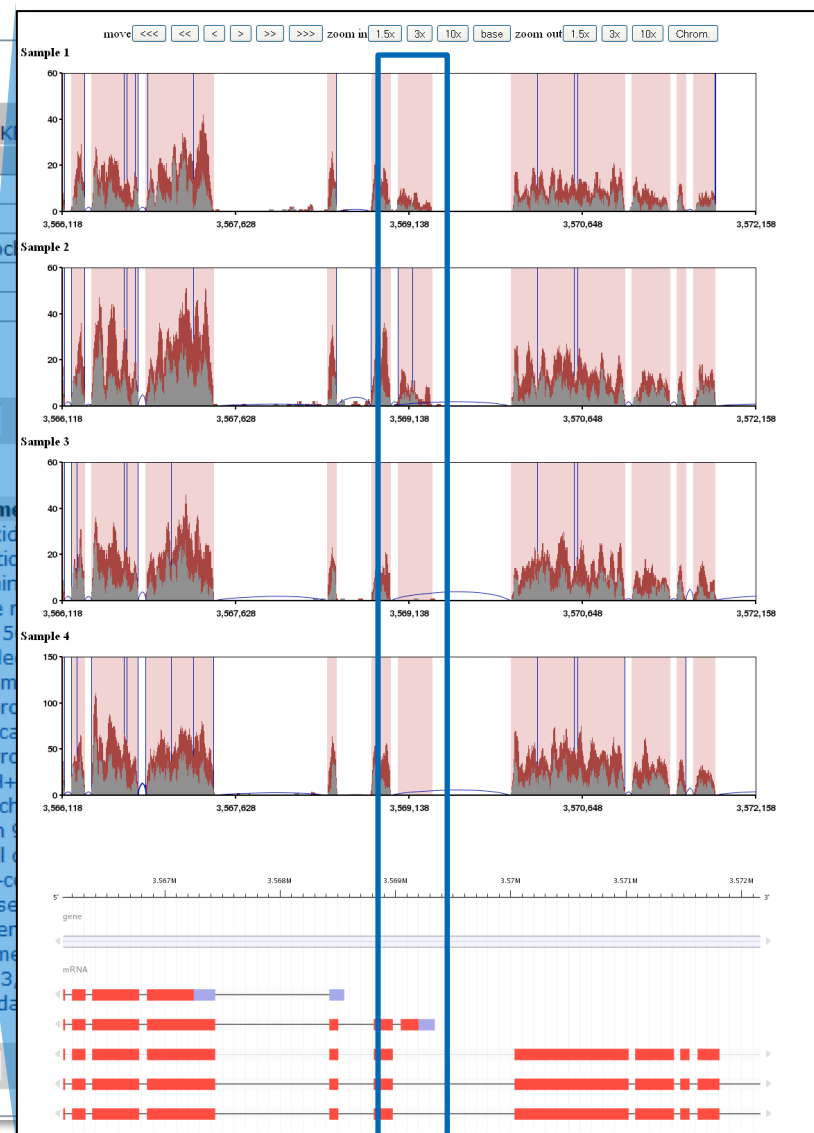
Quality Cutoff: 5

Data Transformation: None

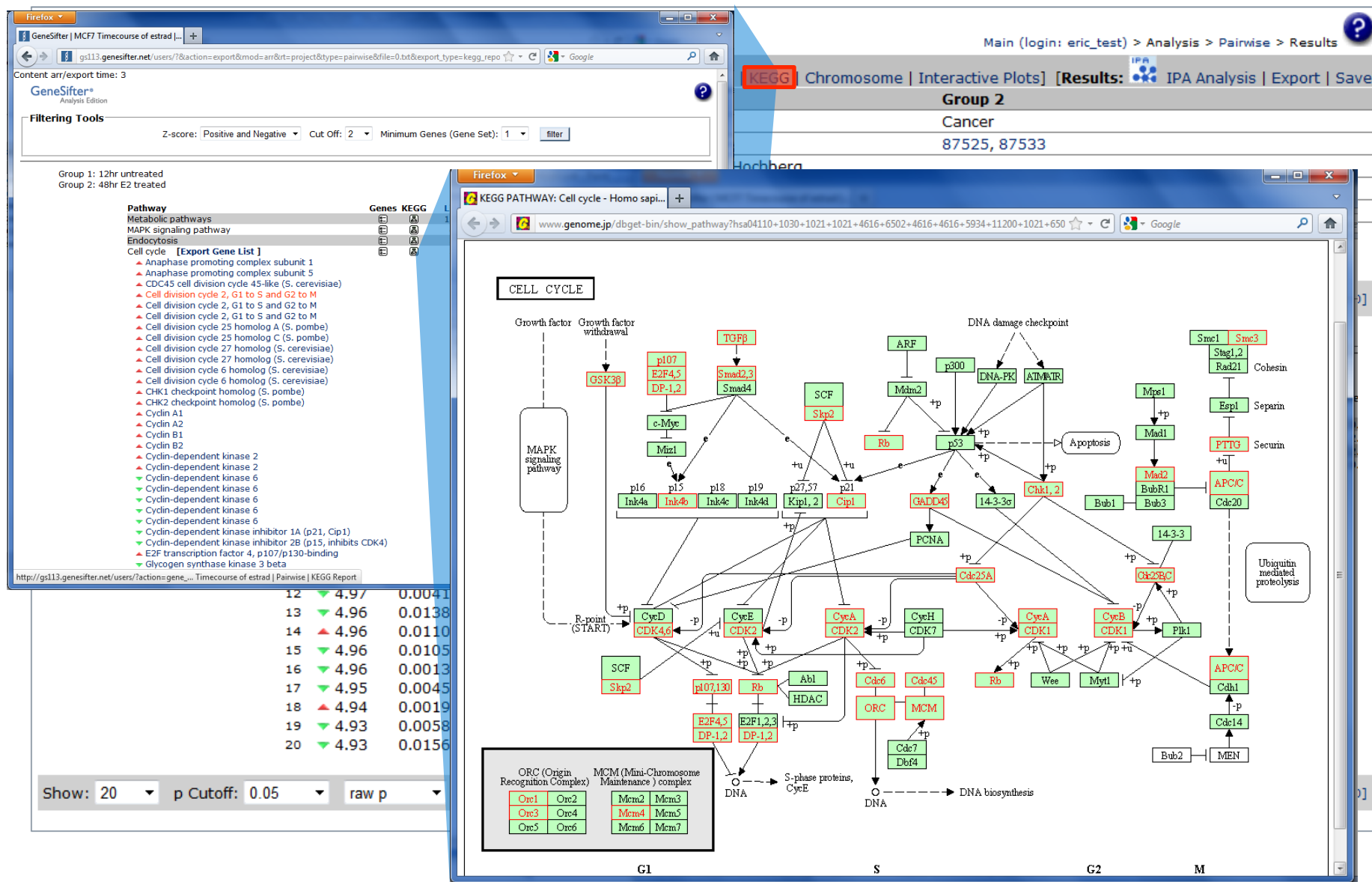
Show: 20 p Cutoff: 0.05 raw p Fold Change: 1.5 Upper: 5

No.	Ratio	p-value	adj. p	Identifier	Gene Name
1	5.00	0.00995	0.22520	NOXO1	NADPH oxidase
2	5.00	0.00168	0.10977	NKD2	Naked cuticle
3	5.00	0.00406	0.15943	TEAD4	TEA domain
4	4.99	0.00410	0.16005	DRD4	Dopamine receptor
5	4.99	0.00678	0.19776	693153	microRNA 5
6	4.99	0.00841	0.21368	SNORD22	Small nucleolar
7	4.99	0.01309	0.25318	TRIM50	Tripartite motif
8	4.98	0.00616	0.18936	DOK3	Docking protein
9	4.98	0.00704	0.19956	100190940	hypothetical
10	4.98	0.02006	0.29120	PTPN20A	Protein tyrosine
11	4.97	0.03240	0.33877	ATP6V0A4	ATPase, H+
12	4.97	0.00419	0.16153	LINGO3	Leucine rich
13	4.96	0.01384	0.25751	AQP9	Aquaporin 9
14	4.96	0.01105	0.23526	CCDC138	Coiled-coil
15	4.96	0.01052	0.22893	GPR25	G protein-coupled
16	4.96	0.00130	0.10153	POLD1	Polymerase delta
17	4.95	0.00454	0.16602	CFD	Complement
18	4.94	0.00198	0.11731	RAB27B	RAB27B, member
19	4.93	0.00587	0.18458	CDH3	Cadherin 3
20	4.93	0.01562	0.26742	SLA	Src-like-adenosine

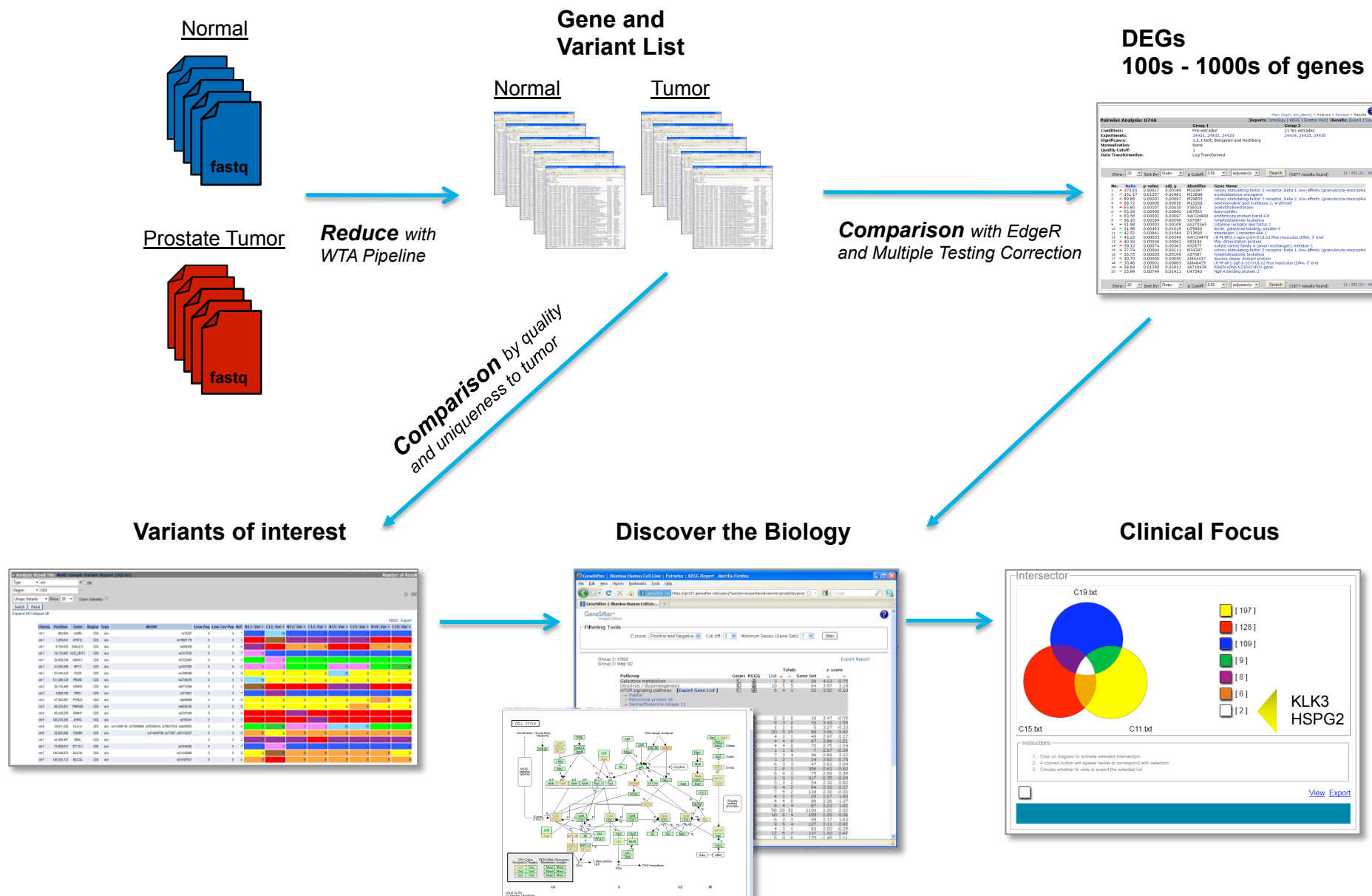
Show: 20 p Cutoff: 0.05 raw p Fold Change: 1.5 Upper: 5



Discover Complex Relationships

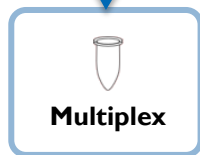


Reduce, Compare, Discover

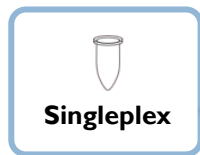


Focus on the Process: GeneSifter Lab Edition

Track samples, complex manipulations



or



GeneSifter Slide Builder

GeneSifter Lab Edition My GeneSifter

Home Orders Lab Data System

Reports

- View Run Reports
- View Instrument Reports

Template Tracker

- Add Template
- Add Templates (Bulk)
- View Templates
- View Template Worksets
- Set up Template Workflows
- Set up Template Types

Primer Tracker

- Add Primer
- View Primers
- Set up Primer Workflow

Primer Mix Tracker

- Add Primer Mix
- View Primer Mixes
- Set up Primer Mix Workflow

Reactions

- Add Reaction Set
- View Reaction Sets
- View Reactions
- Set up Reaction Types

Reaction Plates

- Add Reaction Plate
- View Reaction Plates

Container Management

- Set up Container Workflows
- Set up Container Types
- Set up Container Classes

Sample Prep

- View Flowcells
- View Slides
- View Picotiter Plates

Instrument Runs

- Add 96 Well Plate Run
- Add 384 Well Plate Run
- Add Illumina Sequencing Run
- Add SOLiD Run
- Add 454 Run
- View Runs
- View Next Gen Run Data

Lab Setup

- Chemistry Rules
- Upload Location Map
- Containers
- Instruments

Add Flowcell

Name: smith_lab_001

Flowcell ID: 234324FE3

Freezer: 16-A

Rack: 35

Bin: 12E

Description: Flowcell for Smith Lab - RNA project #1

Assigned To: fejiwfe38832@geospi

Priority: High

Selected Lane:

Lane	n/a
Reaction	human_RNA_03
Workset (Multiplex)	multiplex_1

Pick Samples

Pick Workset

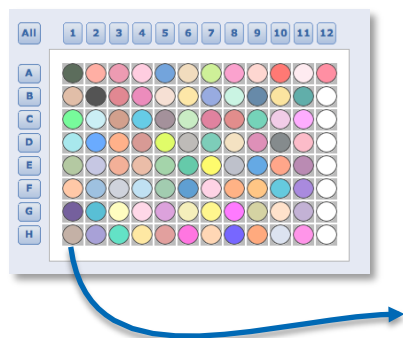
Empty Selected

Deselect All

Create

collect, sort, and deliver

View Sample Details in an Easy to View Format



Template Details

[Next Step](#)
[Edit Template](#)
[Upload Attachment](#)
[Set to Archived](#)
[Delete](#)

Sample QC

1. Qubit Fluorometer
2. LabChip GX

TruSeq Small RNA

1. TruSeq Small RNA Kit Info
2. Ligate 3' Adapter
3. Ligate 5' Adapter
4. Reverse Transcribe and Amplify
5. PCR Amplification
6. Purify cDNA Construct

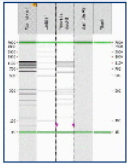
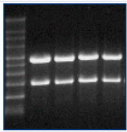
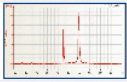
Library Validation

1. Qubit Fluorometer
2. LabChip GX

Name	spleen_597	Template Workset	Cancer_Group_5
Template ID	3621	Parent	n/a
Template Type	Library Validation	Volume (µL)	50
Barcode	79521671	Concentration (ng/µL)	150
Archived	No	Storage Location	Freezer 2
Order Name	n/a	MID	n/a
Description	Created from Template spleen_597		

Related Links: [Details](#), [Workflow History](#), [Results](#), [Relationships](#), [Attachments \(3\)](#)

Template Attachments

Filename	File type	Image	File Size	Upload Step	Field Label	Uploaded By	Uploaded At
LabChip_GX_1.png	png file		135.9 kB	LabChip GX	LabChip File	LabManager	2012-01-24 14:11:30-08
Gel_Image.jpg	unknown		17.3 kB	Purify cDNA Construct	Gel Image	LabManager	2012-01-24 14:09:42-08
LabChip.png	png file		110.4 kB	LabChip GX	LabChip File	LabManager	2012-01-24 14:07:13-08

Workflow History

Workflow	Workflow Version	State	Step	Updated By	Updated At	Comment	Info
Library Validation	28-68058228	Ready	LabChip GX	LabManager	2012-01-24 14:11:30-08	n/a	LabChip File: LabChip_GX_1.png LabChip GX: --Select Chip--
TruSeq Small RNA	70-68058228	Ready	Purify cDNA Construct	LabManager	2012-01-24 14:09:42-08	n/a	Novex Gel lot #: 089085-12 Gel Image: Gel_Image.jpg
TruSeq Small RNA	70-68058228	Not Ready	Ligate 3' Adapter	LabManager	2012-01-24 14:08:37-08	n/a	Total RNA (µg): 5 Thermal Cycler: Thermal Cycler #2
TruSeq Small RNA	70-68058228	Not Ready	TruSeq Small RNA Kit Info	LabManager	2012-01-24 14:07:56-08	n/a	TruSeq kit lot #: 324234-C
Sample QC	39-68058228	Ready	LabChip GX	LabManager	2012-01-24 14:07:13-08	n/a	LabChip GX: RNA-6000 RIN value: 9 LabChip File: LabChip.png

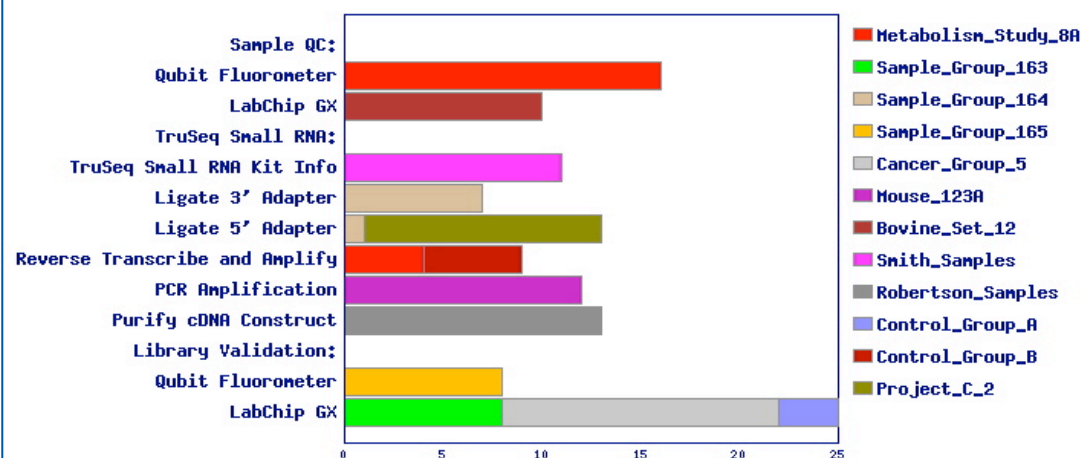
Also includes:
Relationships
Change log
Links to data

To meet
regulatory needs

Know Where Samples Are in the Process

GeneSifter Protocol Viewer

Label	Illumina TruSeq Small RNA - LabChip	Archived	No
Created By	LabManager	Created At	2012-01-24 13:23:20.707987-08
Modified By	LabManager	Modified At	2012-01-24 13:37:04.809804-08
Component Workflows	Sample QC, TruSeq Small RNA, Library Validation		
Description	n/a		



The data browser below shows Templates and Template Worksets that have or need pending work.

Template Worksets (12) Current Templates (124) All Templates					
Find:	Label	CONTAINS		Go	Reset ?
Items 1-12 of 12					
Label	Templates	Barcode	Storage location	Assigned to	Description
Smith Samples	11	n/a	n/a	n/a	n/a
Sample Group 165	8	n/a	n/a	n/a	n/a
Sample Group 164	8	n/a	n/a	LabManager	n/a
Sample Group 163	8	n/a	n/a	n/a	n/a
Robertson Samples	13	n/a	n/a	n/a	n/a
Project C_2	12	n/a	n/a	n/a	n/a
Mouse 123A	12	n/a	n/a	n/a	n/a
Metabolism Study 8A	20	n/a	n/a	n/a	n/a
Control Group B	5	n/a	n/a	n/a	n/a
Control Group A	3	n/a	n/a	n/a	n/a
Cancer Group 5	14	n/a	n/a	n/a	n/a
Bovine Set 12	10	n/a	n/a	n/a	n/a

Review Data Quality

GeneSifter Run Reports

- Software routines link data to samples
- Vendor reports, and Geospiza reports are linked for review.
- Single-click distribution of lab data to collaborators

Instrument Run Results

Rebuild Quality Reports Edit Results Path

Files less than 9.5 GB are linked below.

Name	100799	Flowcell ID	12345
Flowcell	Demo_flowcell	Run ID	145
Run Barcode	145	Base Reaction Type	Illumina Sequencing
State	Completed	Reactions	8
Run Type	Illumina Sequencing	Instrument	HiSeq #1
Completed At	2010-10-25 14:20:32-07		
Results Path	/mnt/nfs/s36/gsl/s47/demo4/ga_index_full		
Description	n/a		

Links: [Run Details](#), [Results \(2023\)](#), [Orders \(1\)](#), [Attachments](#), [Analysis Jobs](#)

Reports: [Results Summary](#), [Intensity Plots](#), [Quality Reports](#)

Sequence Data (8) Run Reports (8) Other Files (2007)

Find: CONTAINS Go Reset ?

Items 1-8 of 8

Filename	File Type	Size	Modified At	Associated Reactions	Order Name	Display to Customer	Analysis Type	Analysis State
s_1_ATCACG_sequence.txt	Text	29.2 MB	2010-09-29 13:40:48	muscle_01	Gregor_Mendel_100799	No	n/a	n/a
s_1_CGATGT_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_02	Gregor_Mendel_100799	No	n/a	n/a
s_1_TGACCA_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_04	Gregor_Mendel_100799	No	n/a	n/a
s_1_TTAGGC_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_03	Gregor_Mendel_100799	No	n/a	n/a
s_2_ATCACG_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_05	Gregor_Mendel_100799	No	n/a	n/a
s_2_CGATGT_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_06	Gregor_Mendel_100799	No	n/a	n/a
s_2_TGACCA_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_08	Gregor_Mendel_100799	No	n/a	n/a
s_2_TTAGGC_sequence.txt	Text	29.2 MB	2010-09-29 13:40:49	muscle_07	Gregor_Mendel_100799	No	n/a	n/a

-- Choose Action -- Go

Run Change Log

Summary	By	At	Comment for Change
Run results path modified.	finch	2010-10-25 16:20:23	n/a
Run results path modified.	finch	2010-10-25 16:20:08	n/a
Run Completed.	LabManager	2010-10-25 14:20:32	n/a
8 Reactions completed as "Run Completed" (C).	LabManager	2010-10-25 14:20:32	n/a
Run downloaded	LabManager	2010-10-25 14:19:08	Run downloaded to instrument HiSeq #1
Run downloaded	LabManager	2010-10-25 14:14:56	Run downloaded to instrument HiSeq #1
Created	LabManager	2010-10-25 14:14:33	n/a

Summary Information For Experiment 070515_SLXA-EAS20_5612 on Machine SLXA-EAS20



Chip Summary

Machine	SLXA-EAS20
Run Folder	070515_SLXA-EAS20_5612
Chip ID	unknown

Lane Parameter Summary

Lane	Sample ID	Sample Target	Sample Type	Length	Filter	Tiles
1	unknown	hg18_refMrna_small	ELAND	36	'((CHASTITY>=0.6))'	Lane 1
2	unknown	hg18_refMrna_small	ELAND	36	'((CHASTITY>=0.6))'	Lane 2
3	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 3
4	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 4
5	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 5
6	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 6
7	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 7
8	unknown	unknown	NONE	unknown	'((CHASTITY>=0.6))'	Lane 8

Tile

Tile	Lane 1	Lane 2
0001		
0002		
0003		
0004		
0005		
0006		

NT Composition

Average QV per Base +/- One STDev

Per Base QV Range

Low QV Analysis

Lane 1

Lane 2

[s_1_sequence.txt](#)

[s_2_sequence.txt](#)

